REN gene

renin

Normal Function

The *REN* gene provides instructions for making a protein called renin, which is produced in the kidneys. This protein is part of the renin-angiotensin system, which regulates blood pressure and the balance of fluids and salts in the body. In the first step of this process, renin converts a protein called angiotensinogen into angiotensin I. Through an additional step, angiotensin I is converted to angiotensin II. Angiotensin II causes blood vessels to narrow (constrict), which results in increased blood pressure. Angiotensin II also stimulates production of the hormone aldosterone, which triggers the absorption of water and salt by the kidneys. The increased amount of fluid in the body also increases blood pressure. Proper blood pressure during fetal growth, which delivers oxygen to the developing tissues, is required for normal development of the kidneys, particularly of structures called the proximal tubules, and other tissues. In addition, angiotensin II may play a more direct role in kidney development, perhaps by affecting growth factors involved in development of kidney structures.

Health Conditions Related to Genetic Changes

REN-related kidney disease

At least four mutations in the *REN* gene have been found to cause *REN*-related kidney disease, a condition in which the kidneys become less able to filter fluids and waste products from the body, resulting in kidney failure. Individuals with this condition have one mutated copy and one normal copy of the *REN* gene in each cell. The mutations involved in *REN*-related kidney disease either change or remove a protein building block (amino acid) in the renin protein. These changes occur in a region of the protein known as the signal sequence, and they impair normal processing of renin. The abnormal protein is toxic to the kidney cells that normally produce renin. The renin-producing cells gradually die off, which disrupts the renin-angiotensin system and causes progressive kidney disease.

renal tubular dysgenesis

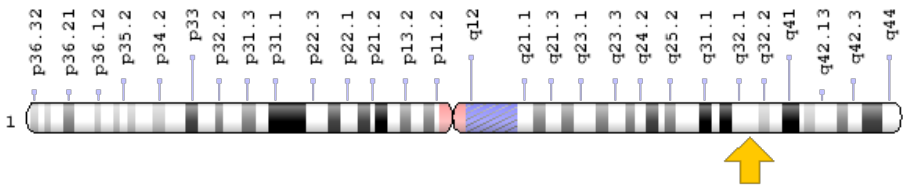
At least 11 mutations in the *REN* gene have been found to cause a severe kidney disorder called renal tubular dysgenesis. This condition is characterized by abnormal kidney development before birth, the inability to produce urine (anuria), and severe low blood pressure (hypotension). These problems result in a reduction of amniotic fluid (oligohydramnios), which leads to a set of birth defects known as the Potter sequence.

Renal tubular dysgenesis can be caused by mutations in both copies of any of the genes involved in the renin-angiotensin system. Most *REN* gene mutations that cause this disorder prevent the production of any renin protein, which results in a nonfunctional renin-angiotensin system. Without this system, the kidneys cannot control blood pressure. Because of low blood pressure, the flow of blood is reduced (hypoperfusion), and the body does not get enough oxygen during fetal development. As a result, kidney development is impaired, leading to the features of renal tubular dysgenesis.

Chromosomal Location

Cytogenetic Location: 1q32.1, which is the long (q) arm of chromosome 1 at position 32.1

Molecular Location: base pairs 204,154,816 to 204,166,337 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- angiotensin-forming enzyme
- angiotensinogenase
- FLJ10761
- HNFJ2
- RENI_HUMAN
- renin precursor, renal
- renin preproprotein

Additional Information & Resources

Educational Resources

- Merck Manual Consumer Version: The Body's Control of Blood Pressure http://www.merckmanuals.com/home/heart-and-blood-vessel-disorders/high-blood-pressure/high-blood-pressure

GeneReviews

- Autosomal Dominant Tubulointerstitial Kidney Disease, REN-Related
  https://www.ncbi.nlm.nih.gov/books/NBK53700

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28REN%5BTI%5D%29+OR+%28renin%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

OMIM

- RENIN
  http://omim.org/entry/179820

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_REN.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=REN%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9958
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5972
- UniProt
  http://www.uniprot.org/uniprot/P00797

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